



# **HL7 Clinical Genomics – From Research to Healthcare**

***NCI/NCRI Joint Conference  
June 13-14, 2011***

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***HL7 Clinical Genomics WG  
Co-chair and Modeling Facilitator***

***HL7 Structured Documents WG  
CDA R2 Co-editor  
CCD Implementation Guide Co-editor***

# Agenda

- **HL7 Clinical Genomics**
- **New Specifications**
- **Experimental Implementations**

# The Mission of HL7 Clinical Genomics Work Group

- The HL7 Clinical Genomics Work Group (CGWG) supports the HL7 mission to create and promote its standards by enabling the communication between interested parties of clinical and genomic data related to an individual. One of the CGWG efforts is the personalization of genomic data – so-called 'omics differences in an individual's genome and its association with relevant phenotypic and clinical information. Associations to investigative/expected phenotypes will be modeled as knowledge that can be leveraged to transform an individual's data into meaningful information.
- CGWG will facilitate the development of common standards for clinical research information management across a variety of organizations -- including national and international government agencies and regulatory bodies, private industry, and sponsored research -- and thus the availability of safe and effective therapies by improving the processes and efficiencies associated with regulated clinical research.
- CGWG will strive to achieve common semantics across clinical and research environments. Consequently, each standardization effort is a building block that later on can be refined to specific real-world applications.



**Healthcare**

**Research**

**Common Semantics**



# Overview of Activities

## Three Tracks:

### v3:

- Family History (Pedigree) Topic
- Genetic Variations Topic
- Gene Expression Topic
- CMETs defined by the Domain

### v2:

#### v2 Implementation Guides




\* The IG “Genetic Test Result Reporting to EHR” is modeled after the HL7 Version 2.5.1 Implementation Guide: Orders And Observations; Interoperable Laboratory Result Reporting To EHR (US Realm), Release 1

### CDA:

- A CDA Implementation Guide for Genetic Testing Reports

### Common:

- Domain Analysis Models for the various topics
- A Domain Information Model (v3) describing the common semantics
- Semantic *alignment among the various specs*

-  Normative
-  DSTU
-  Informative

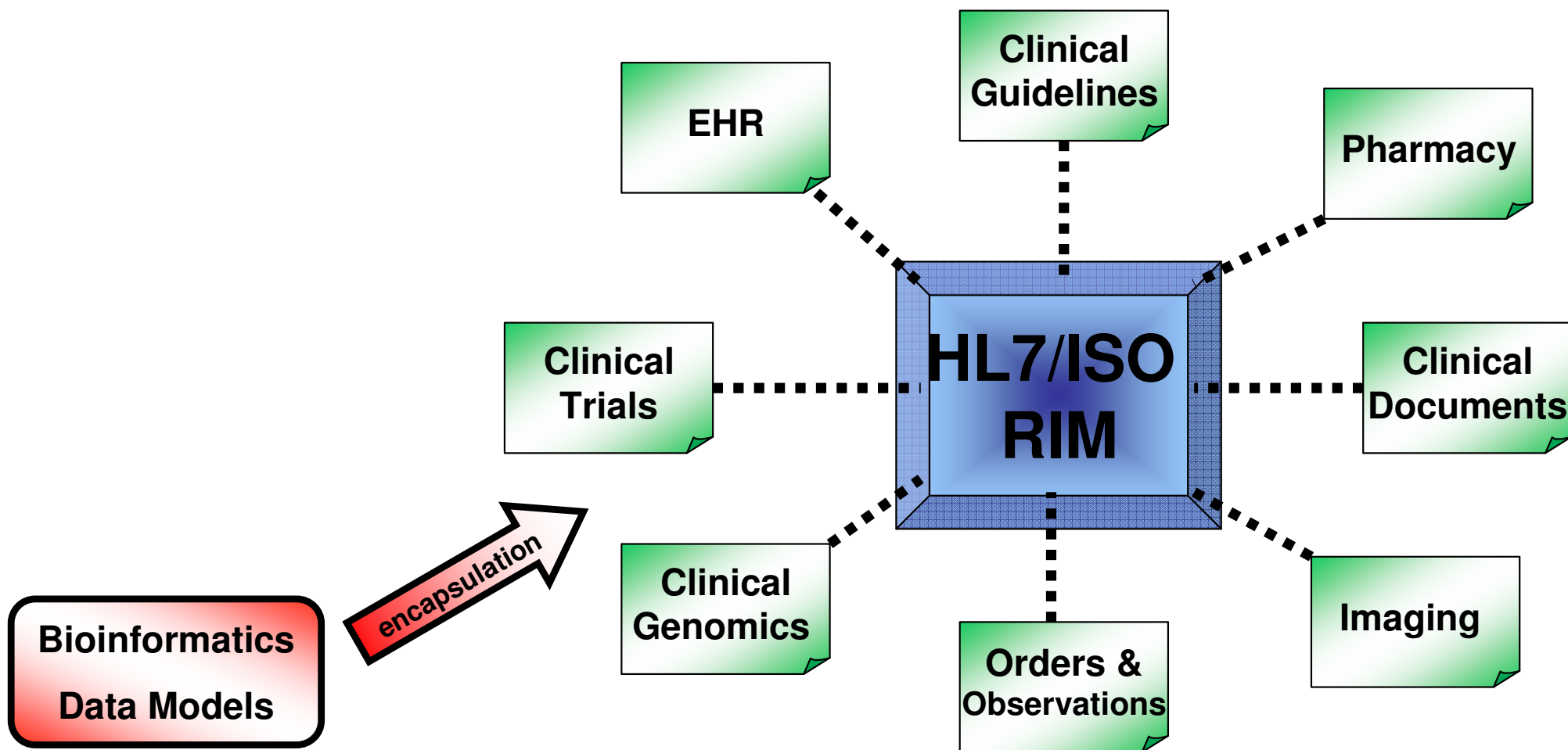


# Main Principles

- **Underlying Model**
  - HL7/ISO Reference Information Model
- **Clinical Genomics Statement**
  - Standard grammar of genotype-phenotype associations
- **Raw genomic patient data**
  - Encapsulate and Bubble Up (through bioinformatics formats)
- **Domain Information Model**
  - The Genome model – Overarching locus and non-locus data
- **Genomics to EHR Systems**
  - CEN EHR 13606 over HL7 RIM
  - Specific Clinical Genomics Statements as:
    - DCMs or Archetypes, over the Clinical Statement model

# To achieve semantic interoperability...

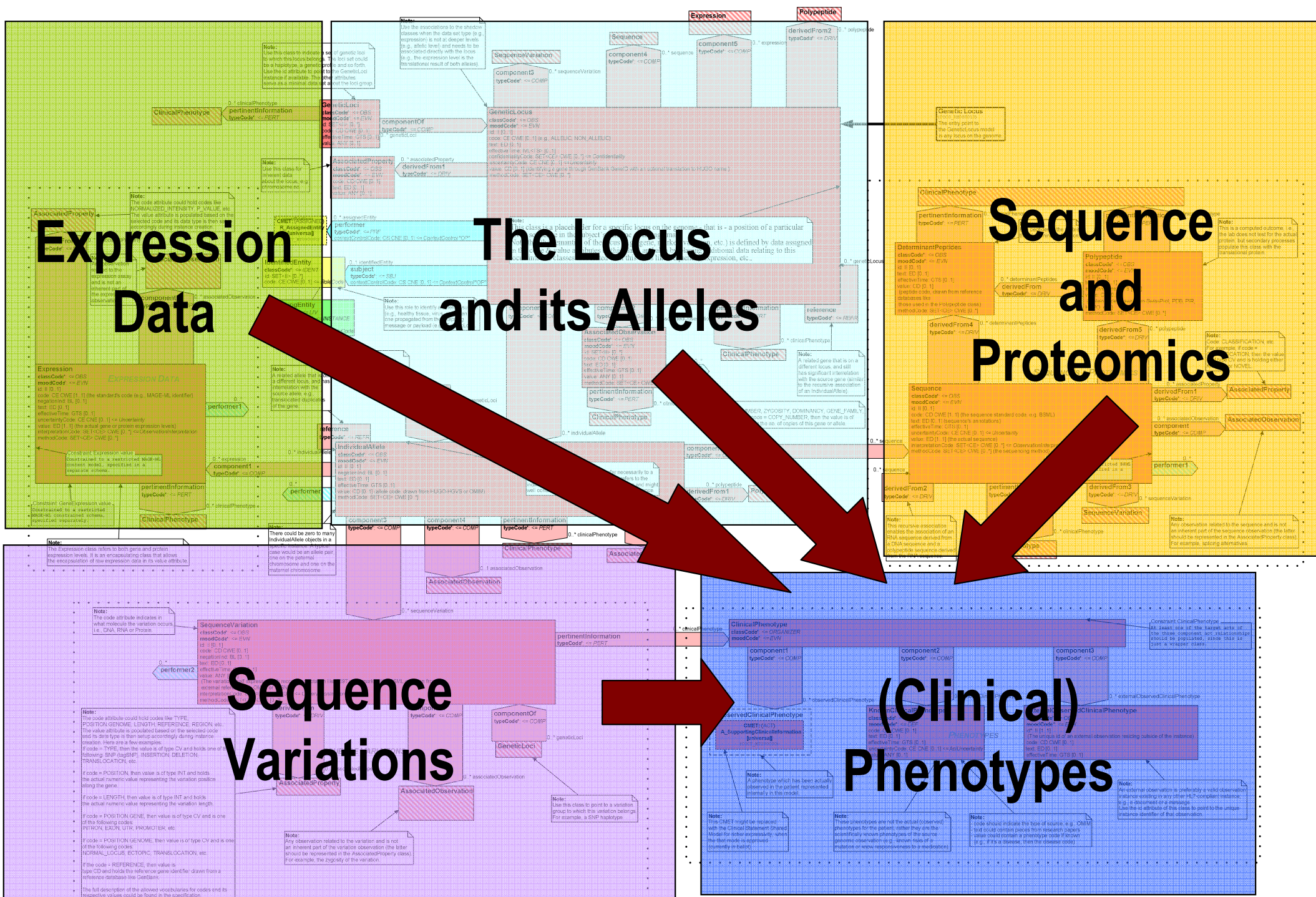
...we need standard specs derived from a reference information model:







# The DSTU GeneticLocus Model Focal Areas:



# The Phenotype Model

## HL7 Clinical Genomics SIG

**Document:** *Genotype Topic - The Phenotype Model*  
**Rev:** COCT\_RM340000UV (Phenotype-v13) **Date:** March 16, 2008  
**Facilitator:** Amnon Shabo (Shvo), IBM Research in Haifa, shabo@il.ibm.com

### Phenotype

(COCT\_RM340000UV)

Entry point to the Phenotype CMET used by the Clinical Genomics models to describe complex phenotypes associated with genomic observations. In case of interpretive phenotypes, this model is used to represent complex phenotypes that cannot be represented by a single code in the interpretationCode attribute of the source genomic observation.

#### Note:

Use this class to reference a phenotype that has been actually observed in the patient and is detailed elsewhere, e.g., in patient medical records.

The attributes hold data replicated from the referenced phenotype and thus can be seen as its metadata / essential data.

#### Note:

Use this class to reference a phenotype known in the scientific literature to be associated with the source genomic observation and is detailed elsewhere, e.g., in knowledge bases, reference repositories and ontology's. Note that these phenotypes have not been observed necessarily in the subject and therefore their mood code is DEF (definitional).

### ObservedOrInterpretive

#### ObservedPhenotype

**classCode\***: <= PHN  
**moodCode\***: <= EVN  
**id**: SET<I> [0..\*]  
**code**: CD CWE [0..1] < ActCode  
**negationInd**: BL [0..1]  
**derivationExpr**: ST [0..1]  
**text**: ED [0..1]  
**statusCode**: CS CNE [0..1] < ActStatus  
**effectiveTime**: GTS [0..1]  
**confidentialityCode**: SET<CE> CWE [0..\*] < Confidentiality  
**uncertaintyCode**: CE CNE [0..1] < ActUncertainty  
**value**: ANY CWE [0..1] < ObservationValue  
**methodCode**: SET<CE> CWE [0..\*] < ObservationMethod

#### InterpretivePhenotype

**classCode\***: <= PHN  
**moodCode\***: <= DEF  
**id**: SET<I> [0..\*]  
**code**: CD CWE [0..1] < ActCode  
**negationInd**: BL [0..1]  
**derivationExpr**: ST [0..1]  
**text**: ED [0..1]  
**statusCode**: CS CNE [0..1] < ActStatus  
**effectiveTime**: GTS [0..1]  
**confidentialityCode**: SET<CE> CWE [0..\*] < Confidentiality  
**uncertaintyCode**: CE CNE [0..1] < ActUncertainty  
**value**: ANY CWE [0..1] < ObservationValue  
**methodCode**: SET<CE> CWE [0..\*] < ObservationMethod

Observed Phenotype

Interpretive Phenotype

#### Note:

Use this CEMT to describe a complex phenotype (e.g., adverse drug reaction in the context of a specific mutation) that is embedded in instances compliant with this mode and not just referenced. The complex phenotype is either known in the scientific literature as a possible phenotype, or has been actually observed in the patient.

**derivedFrom**  
**typeCode\***: <= DRIV

0..\* supportingClinicalStatement

**CMET: (ACT)**  
**A\_SupportingClinicalStatement**  
**[universal]**  
 (COCT\_MT530000UV)

**patient**  
**type**

\* medication

**CMET: (ADMM)**  
**R\_Medication**  
**[universal]**  
 (COCT\_MT230100UV)

#### Note:

Use this CMET to describe medication components related to this phenotype (e.g., in pharmacogenomics uses), when the administration of the drug is described elsewhere.

**performer**  
**typeCode\***: <= PRF  
**time**: IVL<TS> [0..1]

0..\* assignedEntity

**CMET: (ASSIGNED)**  
**R\_AssignedEntity**  
**[universal]**  
 (COCT\_MT090000UV)

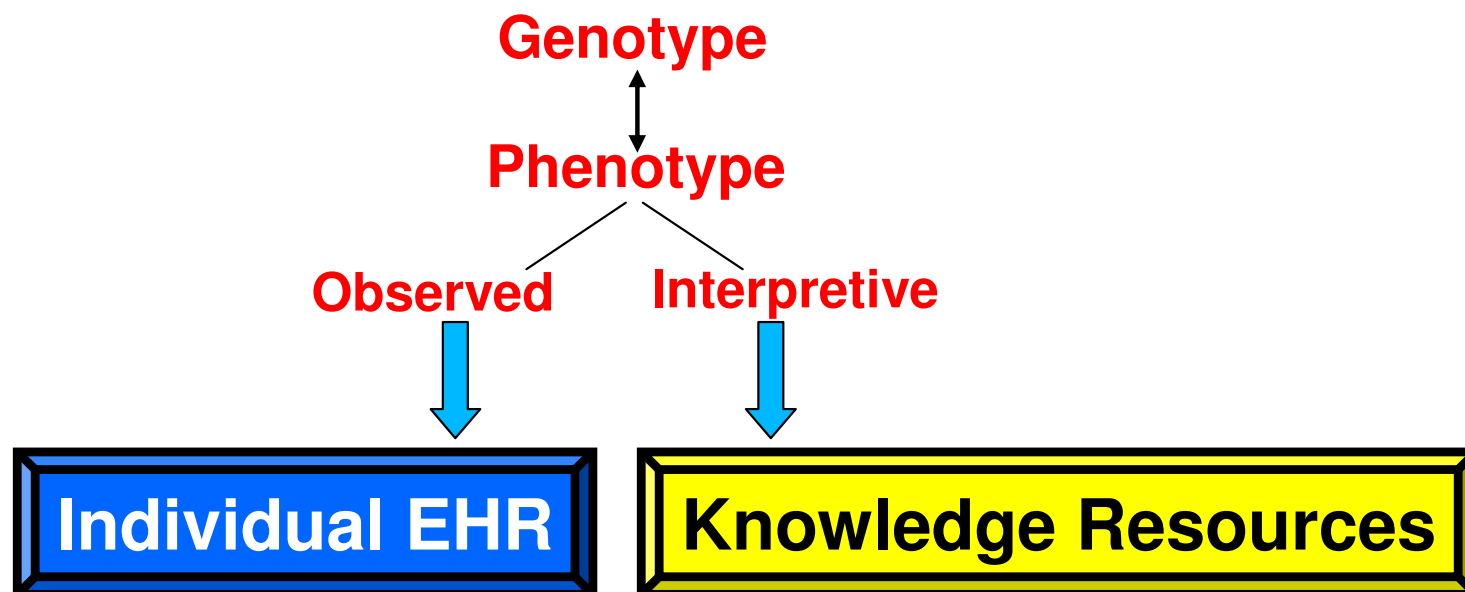
**author**  
**typeCode\***: <= AUT

0..\* assignedEntity



# Genotype-Phenotype Associations

- In clinical environments:
  - Observed versus interpretive phenotypes
  - Observed should reside in the EHR
  - Interpretive should be related to knowledge base



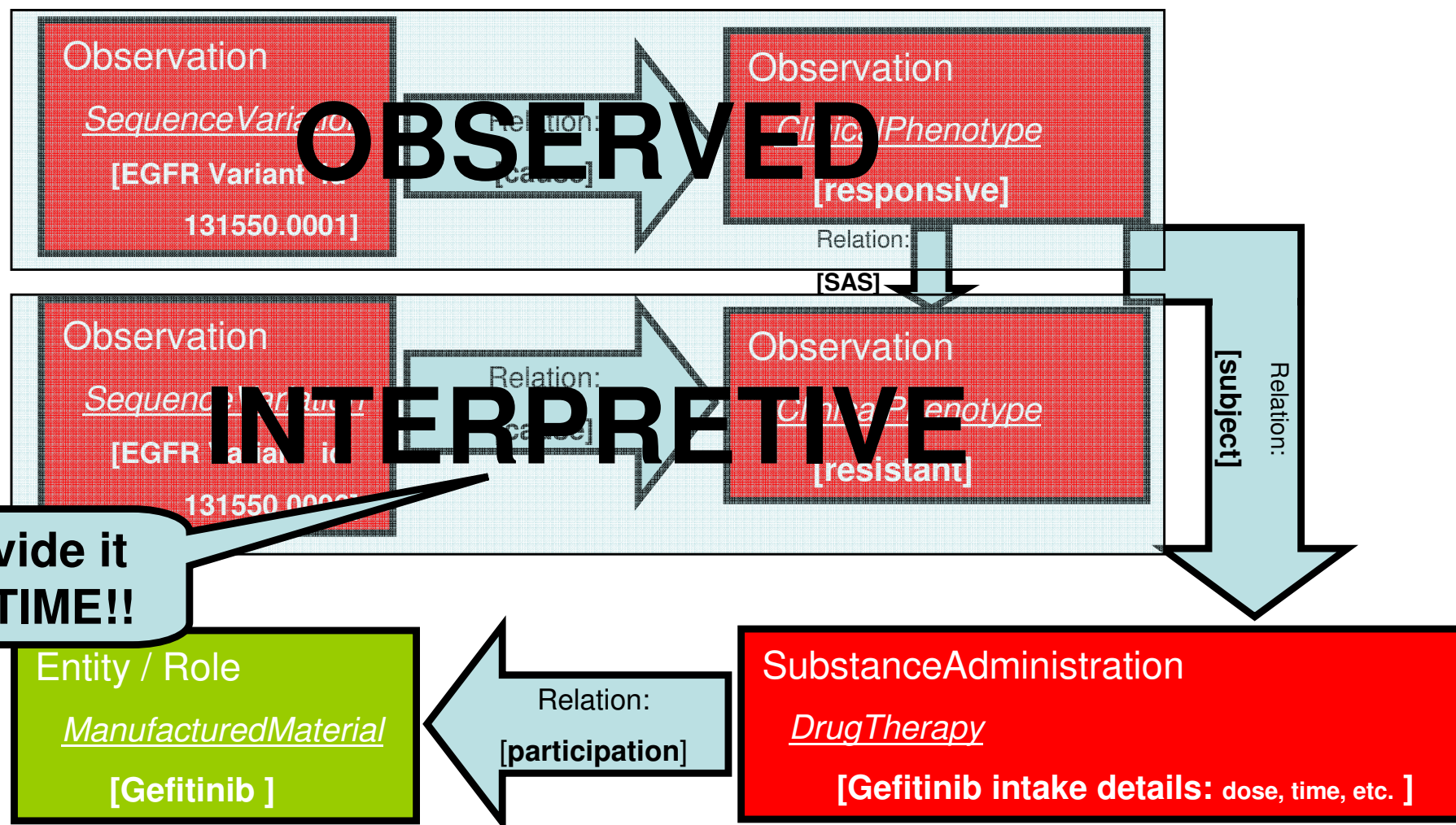


# From Data to Knowledge

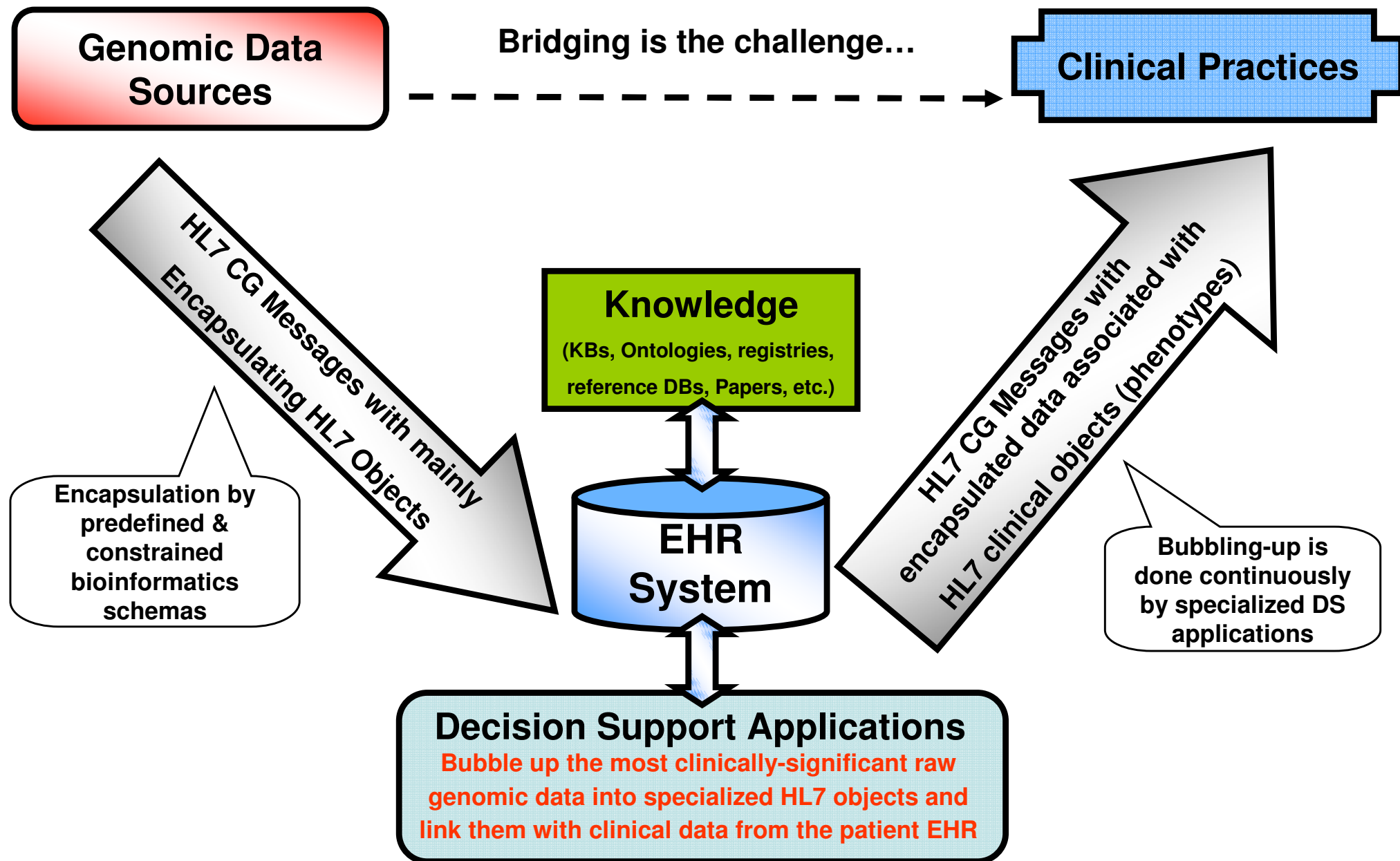
e.g., an OMIM Entry:

*Despite the **dramatic responses** to EGFR inhibitors in patients with non-small cell lung cancer, most patients ultimately have a **relapse**. {12:Kobayashi et al. (2005)} reported a patient with EGFR-mutant, Gefitinib-responsive, advanced non-small cell lung cancer who had a relapse after 2 years of **complete remission** during treatment with Gefitinib. The DNA sequence of the EGFR gene in his tumor biopsy specimen at relapse revealed the presence of a **second mutation** ({131550.0006}). Structural modeling and biochemical studies showed that this second mutation led to the **Gefitinib resistance**.*

# Example: Structuring OMIM Entries (cont.)

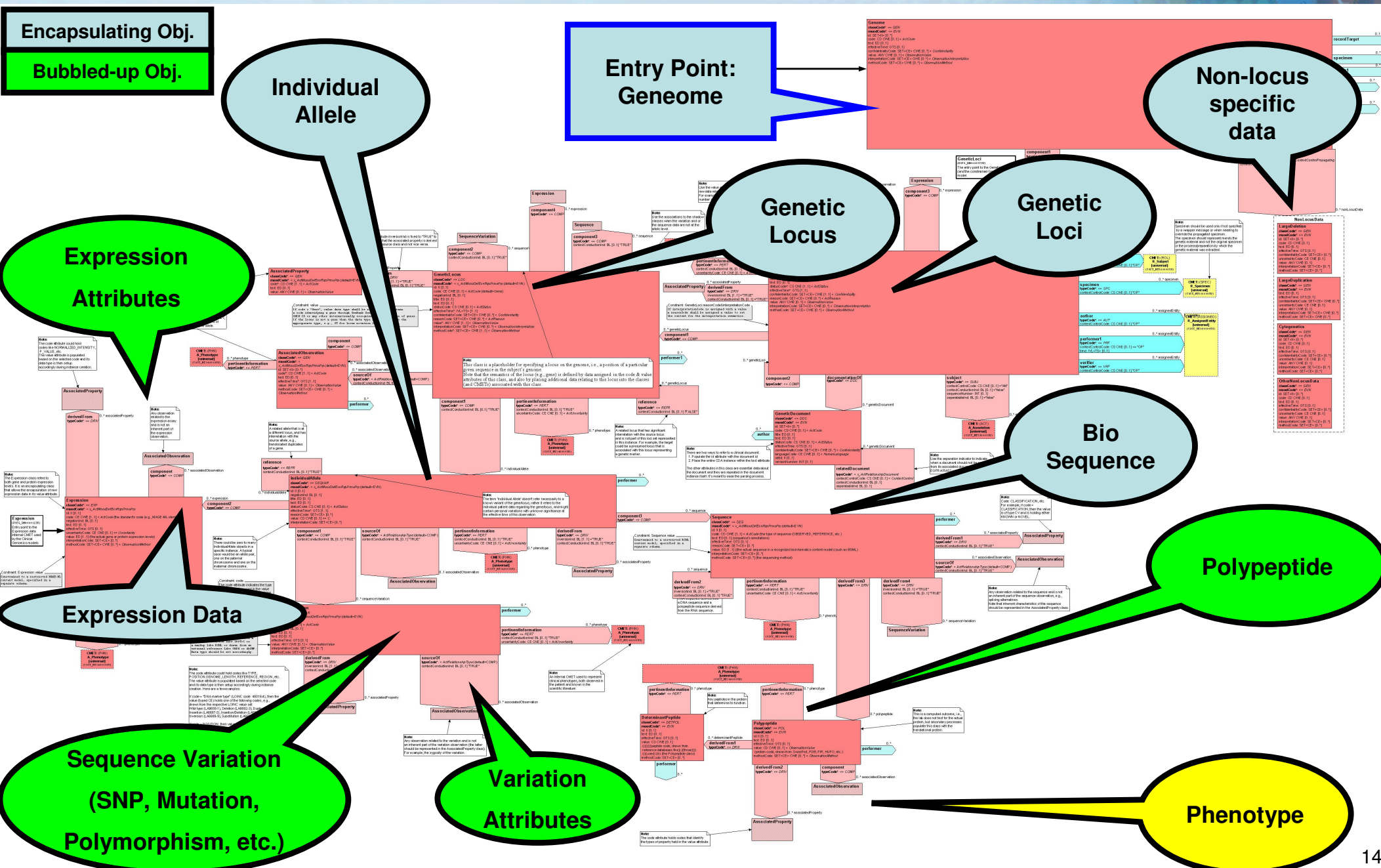


# The Underlying Paradigm: Encapsulate & Bubble-up





# The Domain Information Model - Genome



# Example: Family History XML Encoding

*Taken from a patient pedigree, the portion related to patient's daughter*

```

<!-- DAUGHTER -->
- <relationshipHolder>
  <id extension="555.011" />
  <code code="DAU" />
+ <relationshipHolder>
  <!-- GENOMIC DATA -->
- <subjectOf>
  - <clinicalGenomicChoice>
    - <clinicalGenomicChoiceGenotype>
      - <Genotype>
        - <individualAllele>
          <code code="BRCA1" codeSystem="[insert GenBank OID]"
            codeSystemName="GenBank" />
          <text>Homo sapiens breast and ovarian cancer susceptibility (BRCA1)
            complete cds.</text>
          + <AlleleSequence>
          + <SequenceVariation>
          </individualAllele>
        </Genotype>
      </clinicalGenomicChoiceGenotype>
    </clinicalGenomicChoice>
  </subjectOf>
  <!-- CLINICAL DATA -->
+ <subjectOf>
</relationshipHolder>
<!-- end of DAUGHTER data -->

```

Bubble  
up...

Point  
back...

To  
phenotype  
and beyond....



# XML Fusion: Encapsulation of Raw Genomic Data

HL7 v3 XML

Raw genomic data represented in  
Bioinformatics markup

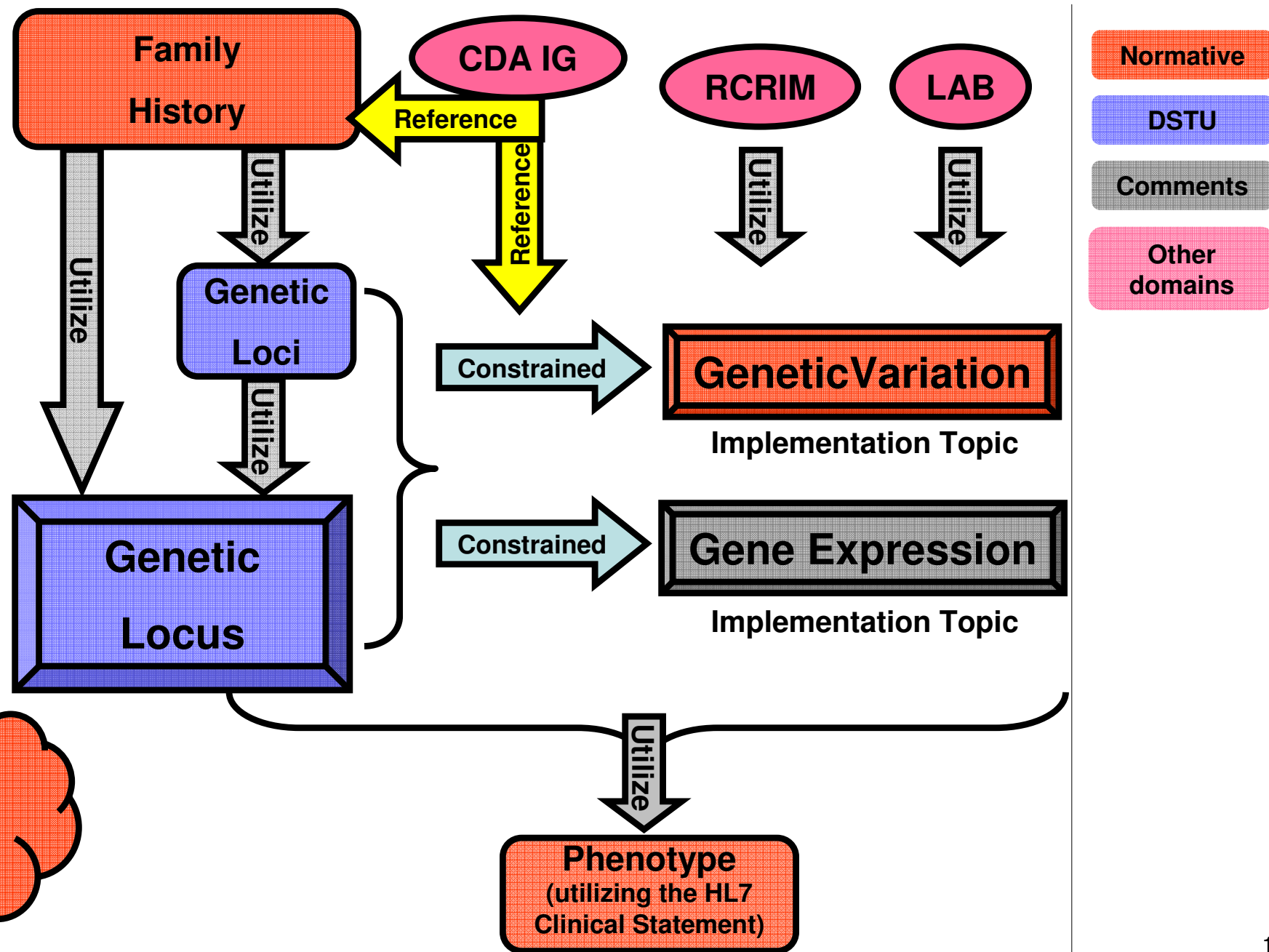
```

<subjectOf2>
  <geneticLocus>
    <component1>
      <individualAllele moodCode="EVN">
        <text>breast cancer 1, early onset</text>
        <value code="83990" displayName="BRCA1" codeSystemName="NCBI Entrez">
          <translation code="20473" displayName="BRCA1" codeSystem="HGNC"/>
        </value>
      <component2>
        <sequence moodCode="EVN">
          <code code="BSMLcon3"/>
          <value mediaType="text/xml">
            <bsml:Bsml xmlns:bsml="urn:bsml.org">
              <bsml:Definitions>
                <bsml:Sequences>
                  <bsml:Sequence id="seq1" molecule="dna" ic-acckey="U14680 REGION: 101..199" db-source="GenBank" title="
BRCA1, exon 2" representation="raw" local-acckey="this could be used by the genetic lab">
                    <bsml:Seq-data>
                      GCTCCCA CTCCATGAGG TATTTCTTCA
                      CATCCGTGTC CCGGCCCGGC CGCGGGGAGC CCCGCTTCAT CGCCGTGGGC
                      TACGTGGACG ACACGCAGTT CGTGCGGTTT GACAGCGACG CCGCGAGCCA
                      GAGGATGGAG CCGCGGGCGC CGTGGATAGA GCAGGAGGGG CCGGAGTATT
                      GGGACCAGGA GACACGGAAT GTGAAGGCC AGTCACAGAC TGACCGAGTG
                      GACCTGGGGA CCCTGCGCGG CTACTACAAC CAGAGCGAGG CCG
                    </bsml:Seq-data>
                  </bsml:Sequence>
                  <bsml:Sequence id="seq2" molecule="dna" ic-acckey="U14680 REGION: 200..253" db-source="GenBank" title="
BRCA1, exon 3" representation="raw" local-acckey="this could be used by the genetic lab">
                    <bsml:Seq-data>
                      GTTCTCA
                      CACCATCCAG ATAATGTATG GCTGCGACGT GGGGTCGGAC GGGCGCTTCC
                      TCCGCGGGTA CCGGCAGGAC GCCTACGACG GCAAGGATTA CATCGCCCTG
                      AACGAGGACC TGCCTCTTG GACCGCGGCG GACATGGCGG CTCAGATCAC
                      CAAGCGCAAG TGGGAGGCGG CCCATGTGGC GGAGCAGCAG AGAGCCTACC
                      TGGATGGCAC GTGCGTGGAG TGGCTCCGCA GATACCTGGA GAACGGGAAG
                      GAGACGCTGC AGCGCACGG
                    </bsml:Seq-data>
                  </bsml:Sequence>
                </bsml:Sequences>
              </bsml:Definitions>
            </bsml:Bsml>
          </value>
        </sequence>
      </component2>
    </individualAllele>
  </component1>
</geneticLocus>
</subjectOf2>

```



# HL7 Clinical Genomics v3 Static Models





# New Specifications under Ballot

# CDA IG for Genetic Testing Reports

## ■ Scope

- Define a universal implementation guide for genetic testing reports that are both human readable and machine-processable

## ■ Design principles

- Follow existing report formats commonly used in healthcare & research
- Emphasize interpretations & recommendations
- Provide general background information on tests performed
- Represent interpretation by utilizing patterns of 'genotype-phenotype' associations in the HL7 v3 Clinical Genomics and implement them as harmonized clinical statement entry-level templates in this IG
- Reference HL7 Clinical Genomics instances as the place holders of raw data (personal evidences), similarly to referencing images (technically-wise)

## ■ CDA Template Editor:

- Developed using the MDHT open source tool (OHT)



# GTR Rendered – The Header

Hearing Loss: Connexin 26 and 30 Full Gene Sequencing Panel Test Report - Windows Internet Explorer

D:\Amnon-eHealth\CDA\CDA Implementation Guides\GTR - Genetic Testing Report\CDA-GeneticTestingReport-Sample-v7.xml

File Edit View Favorites Tools Help

★ Favorites TeamForge : Project Home Hearing Loss: Connexin 2... X

## Hearing Loss: Connexin 26 and 30 Full Gene Sequencing Panel Test Report

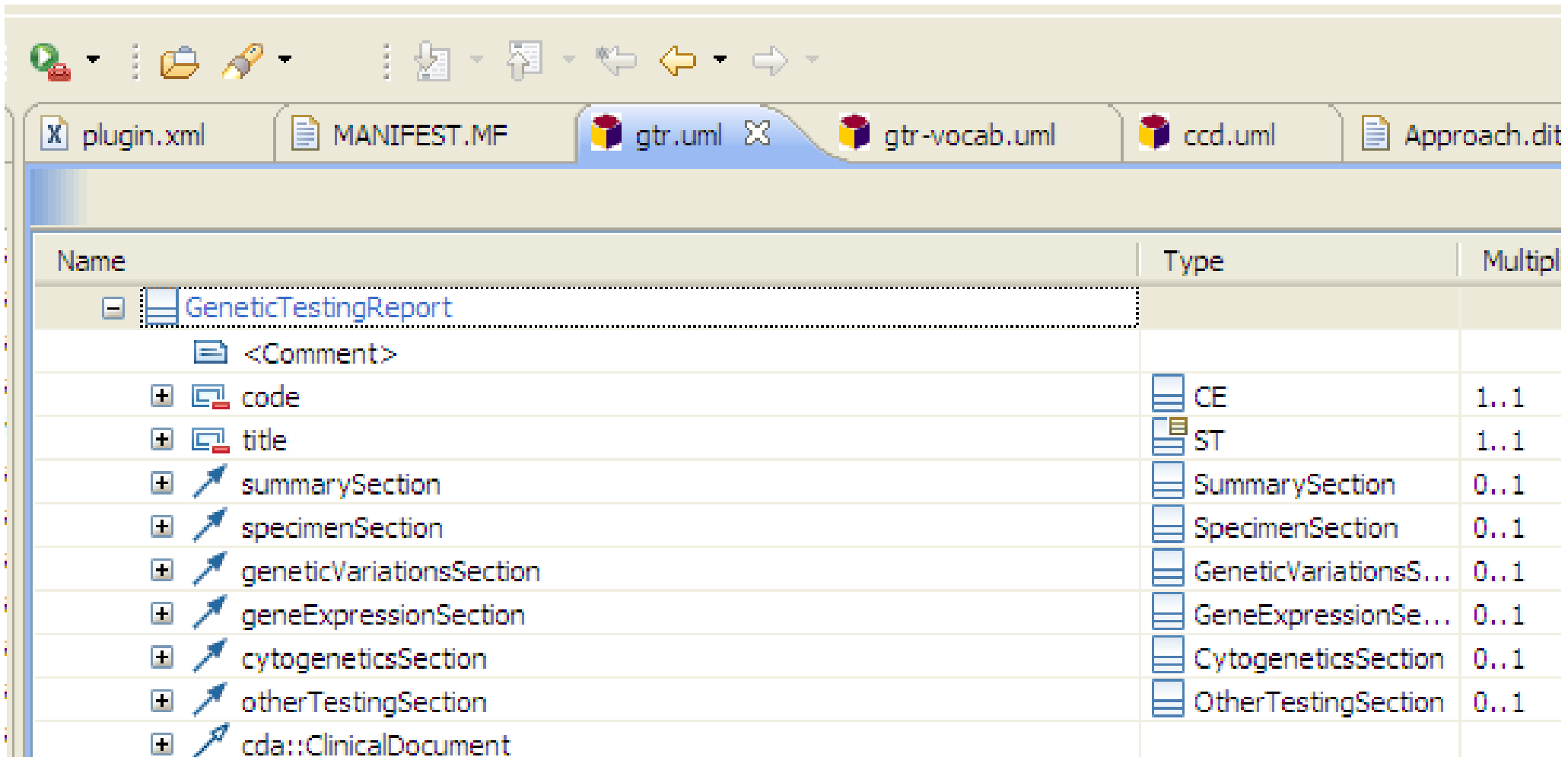
|                               |   |                    |  |
|-------------------------------|---|--------------------|--|
| <b>Patient</b>                | John Doe  |                    |  |
| <b>Date of birth</b>          | May 5, 1947   | <b>Sex</b>         | Male                                       |
| <b>Contact info</b>           | address not available<br>Telecom information not available                    | <b>Patient IDs</b> | 123456789 2.16.840.1.113883.18.12.7.30.9.2 |
| <b>Document Id</b>            | c266 2.16.840.1.113883.18.12.7.30.9.1   |                    |  |
| <b>Document Created:</b>      | August 9, 2010  |                    |  |
| <b>Author</b>                 | Jean Genome,  |                    |  |
| <b>Legal authenticator</b>    | Jean Genome of The New Genetic Testing Laboratory signed at February 12, 2006 |                    |  |
| <b>Document maintained by</b> | 2.16.840.1.113883.19.3.2409   |                    |  |

## Table of Contents

- [Summary Section](#)
- [Genetic Variations Section](#)
- [Genetic Variations Section](#)
- [Genetic Variations Section](#)

Draft that has not been clinically validated

# CDA GTR Section Outline

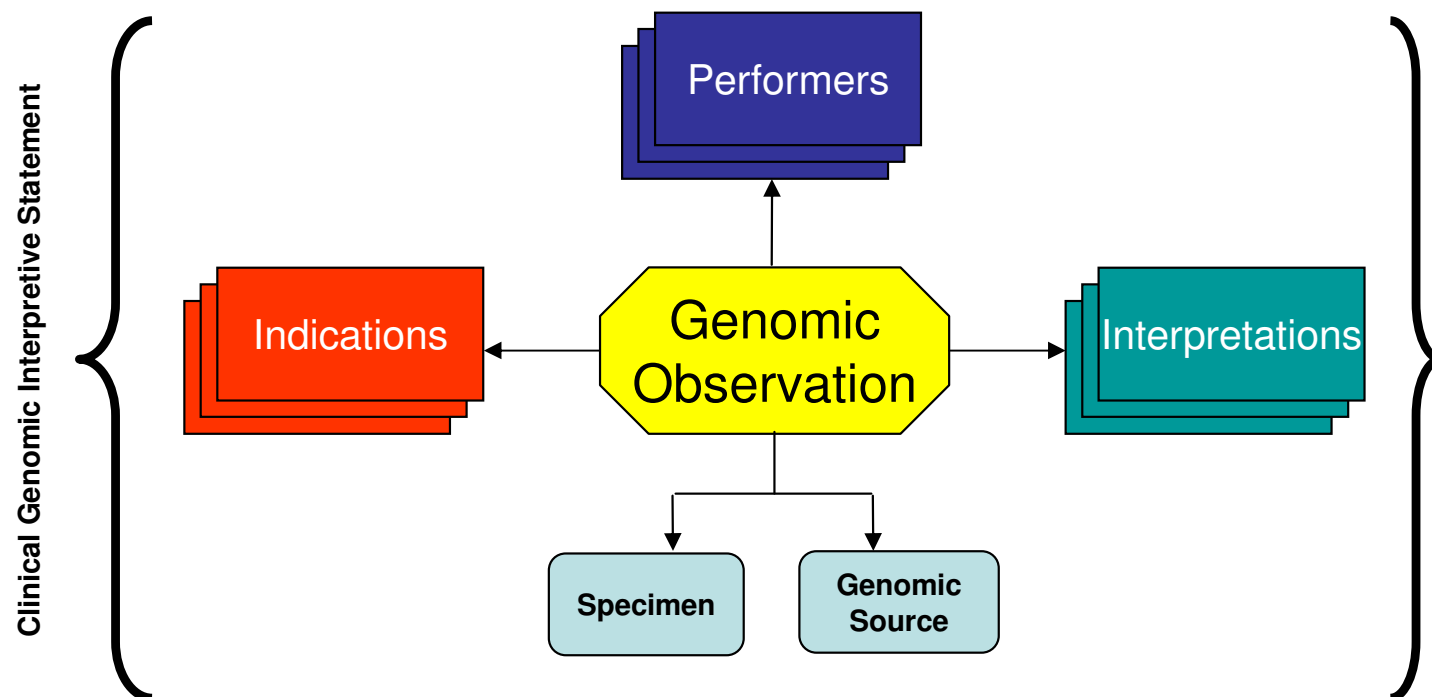


The screenshot shows a UML modeling tool interface with a toolbar at the top and a tabbed workspace. The active tab is 'gtr.uml'. Below the tabs, a table displays the structure of the 'GeneticTestingReport' class. The table has three columns: 'Name', 'Type', and 'Multiplicity'. The 'GeneticTestingReport' class is expanded, showing its components: a comment, a code element, a title element, and several sections (summarySection, specimenSection, geneticVariationsSection, geneExpressionSection, cytogeneticsSection, otherTestingSection) and a base class 'cda::ClinicalDocument'.

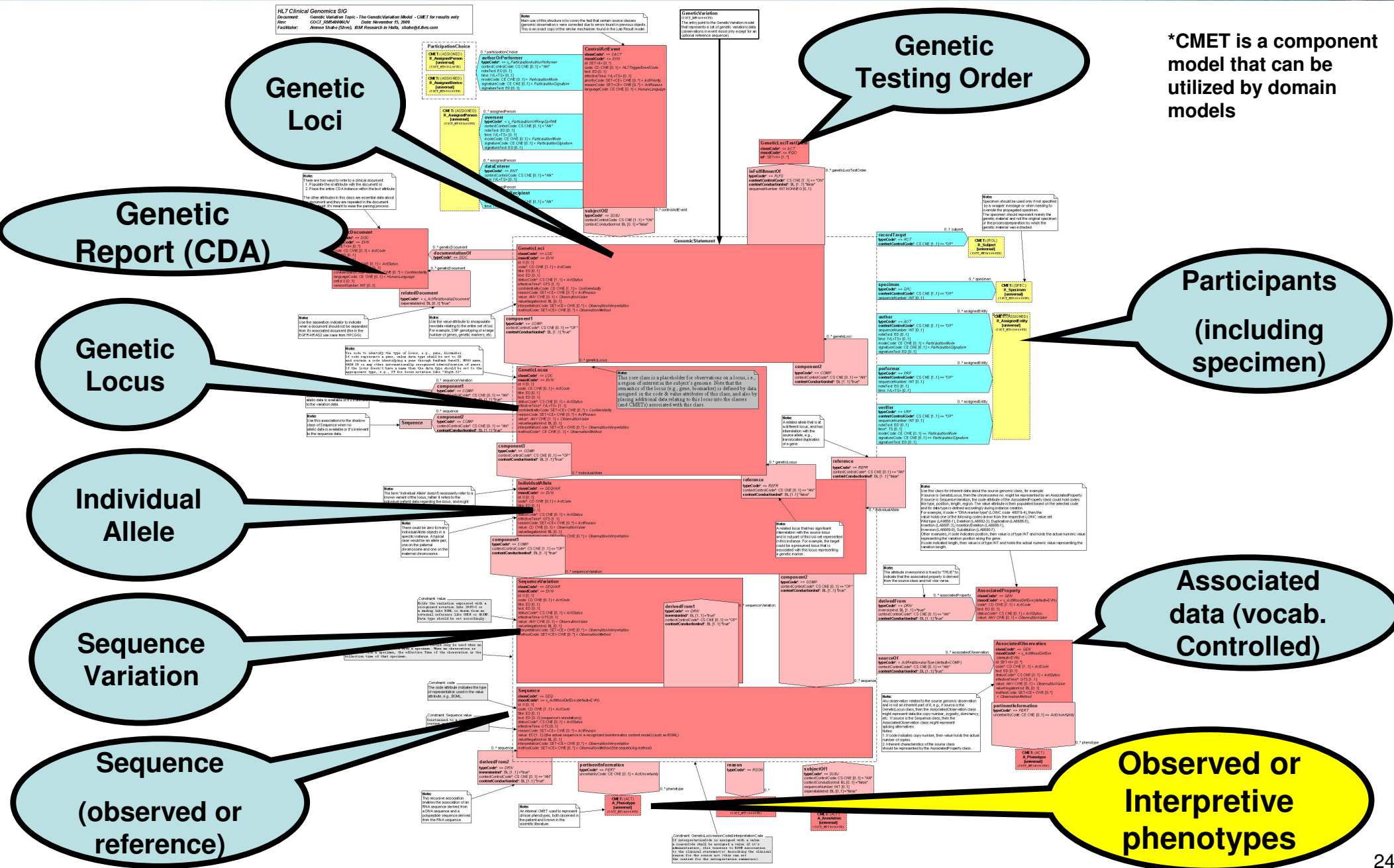
| Name                     | Type                  | Multiplicity |
|--------------------------|-----------------------|--------------|
| GeneticTestingReport     |                       |              |
| <Comment>                |                       |              |
| code                     | CE                    | 1..1         |
| title                    | ST                    | 1..1         |
| summarySection           | SummarySection        | 0..1         |
| specimenSection          | SpecimenSection       | 0..1         |
| geneticVariationsSection | GeneticVariationsS... | 0..1         |
| geneExpressionSection    | GeneExpressionSe...   | 0..1         |
| cytogeneticsSection      | CytogeneticsSection   | 0..1         |
| otherTestingSection      | OtherTestingSection   | 0..1         |
| cda::ClinicalDocument    |                       |              |

# The Clinical Genomic Statement

- An abstract Clinical Genomic Statement (CGS) template that
  - has at its core a genomic observation (e.g., a DNA sequence variation)
  - If it's a major observation, then it should be associated with indications and interpretations, specimen and genomic source class, and optionally associated with performers
  - If it's an associated observation (e.g., amino acid change), then only the genomic observation is populated and optionally the performers
- The CGS abstract template is instantiated by specialized CGS's, e.g., for genetic variations or cytogenetics, as well as for their associated observation



# The Genetic Variation CMET\* (passed normative in Jan. 2010)

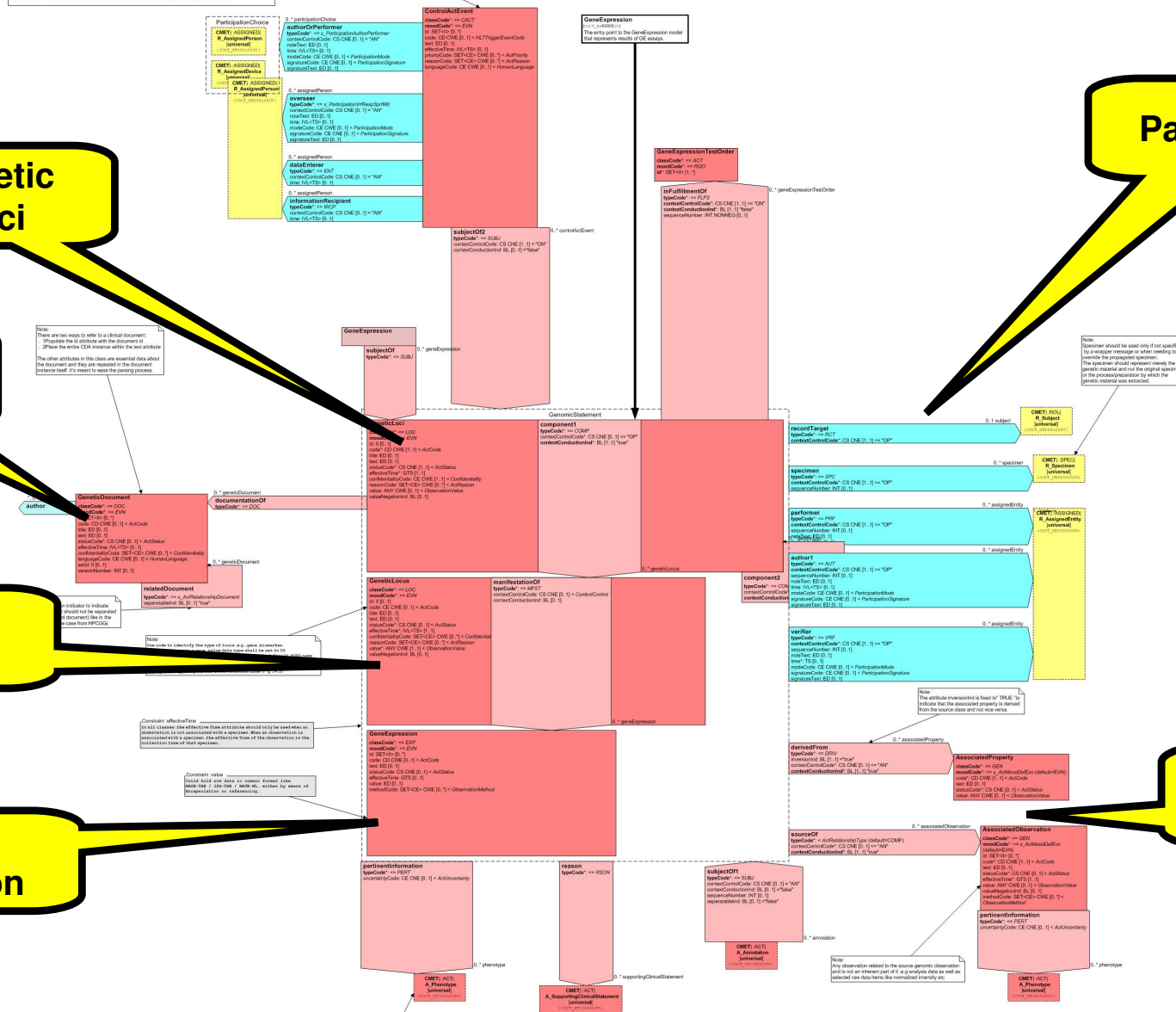




# The Gene Expression CMET Draft

HL7 Clinical Genomics WG  
Document: Genomic Variation Topic - The Gene Expression Model - CMET  
Rev: CDDT\_000000000V Date: September 15, 2010  
Facilitator: Atsushi Shiohara, IBM Research in Haifa, shiohara@il.ibm.com

Note:  
Main use of this structure is to convey the fact that certain source classes  
specimens, information, were collected due to events, such as previous studies.  
This is an exact copy of the similar mechanism found in the Lab Result model.



Participants

Genetic Loci

GTR Report

Genetic Locus

Gene Expression

Associated observations

# Experimental Implementations

- V3 specs
  - The Genetic Variation and Pedigree models are used in Hypergenes (a European project on essential hypertension, <http://www.hypergenes.eu/>)
  - The Family History spec is widely used in (e.g., MGH, HHS)
  - The Pedigree and Genetic Variation models are used by the Rizzoli institute in Bologna, Italy for orthopedic genetic diseases
- CDA GTR
  - Used in Korea in the uHealth project (Gil Hospital)
- v2 IG
  - Used by Harvard and Intermountain to send genetic testing results message

# Hypergenes – Essential Hypertension Genomics

## ■ Challenge & Objectives

- **An EC-FP7 funded project** addressing challenge **HEALTH-2007-2.1.1-2**: Molecular epidemiological studies in existing well characterized European (and/or other) population cohorts
- **Objective**: To define a comprehensive genetic epidemiology **disease model** of **essential hypertension** (EH) by integrating new technologies of high-throughput genotyping with sophisticated statistical-mathematical modeling and methods of genetic epidemiology

## ■ Scientific Coordinator

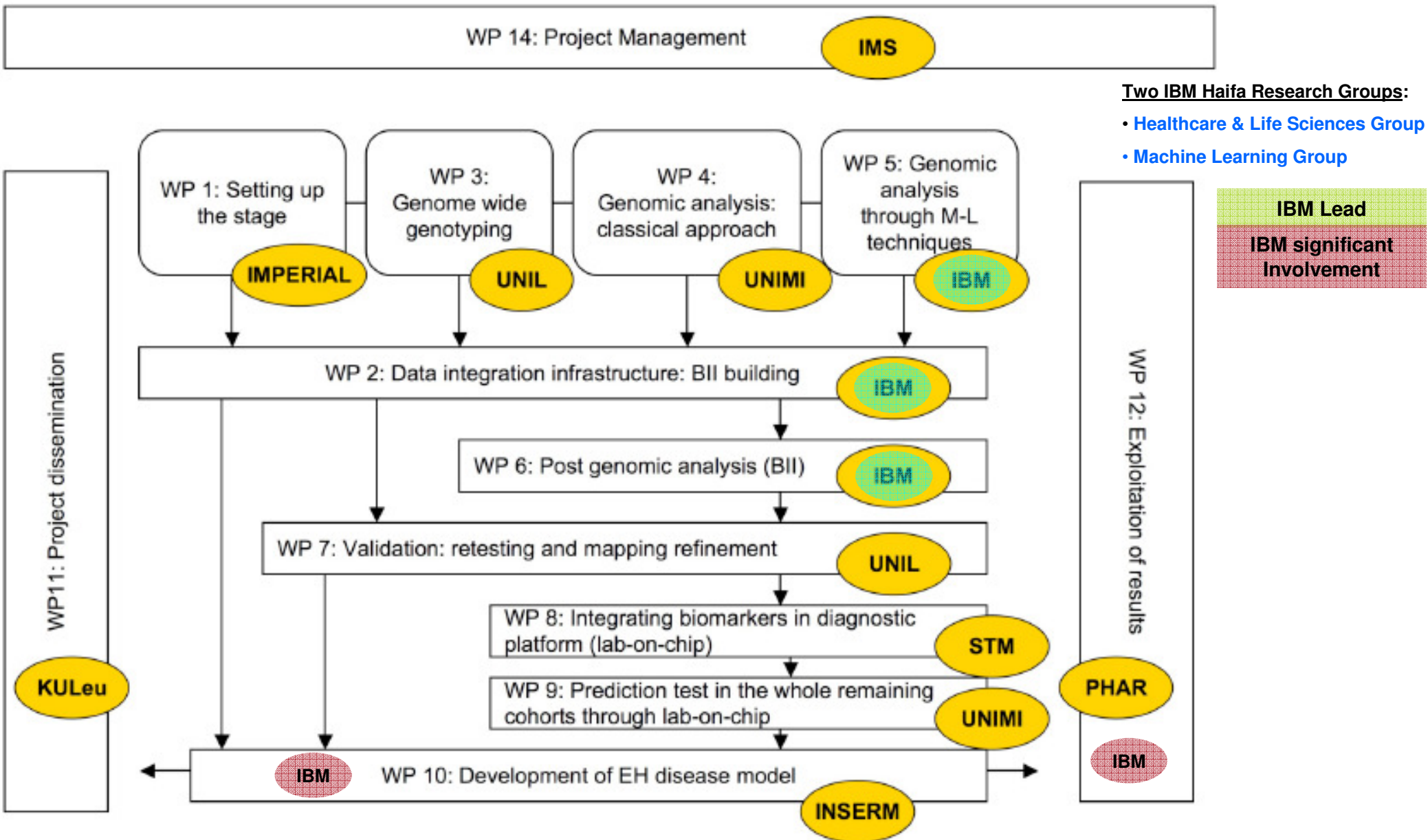
- State University of Milano

## ■ Collaborators

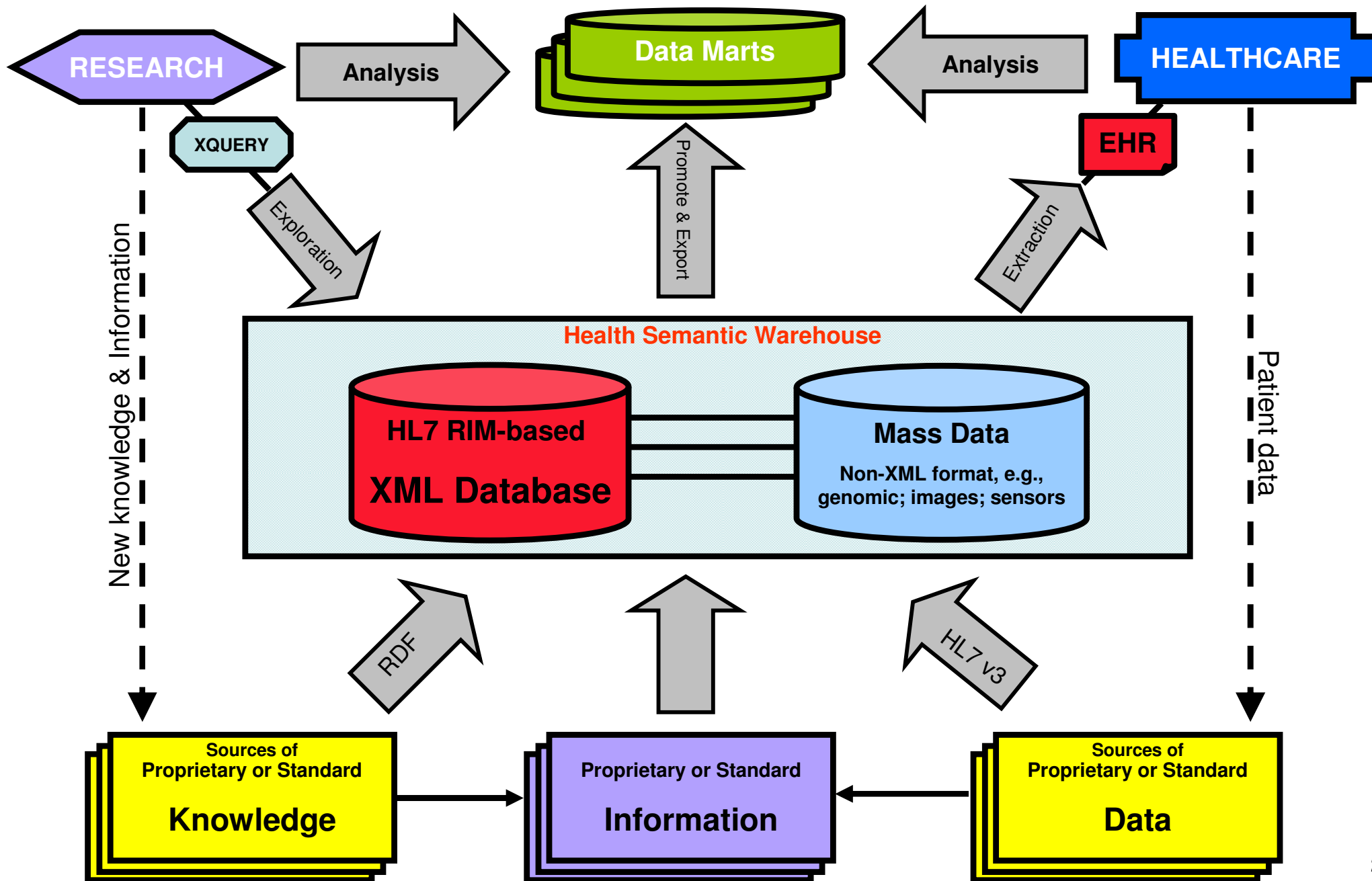
- State University of Milano; Katholieke Universiteit Leuven; Uniwersytet Jagiellonski Collegium Medicum; Sineurra; IMS Research; State Scientific Research Institute of Internal Medicine, Russian Academy of Medical Sciences Siberian Department; Imperial College London; UC San Diego; INSERM - College de France; Warwick Medical School; Prassis-SigmaTau Research Institute, Milano; STMicroelectronics; Losanna & Ginevra University; Pharm-Next; Softeco Sismat Spa, Genoa; Shanghai Institute of Hypertension; Charles University in Prague; Faculty of Medicine in Pilsen; State University of Padova; Medical University of Gdansk



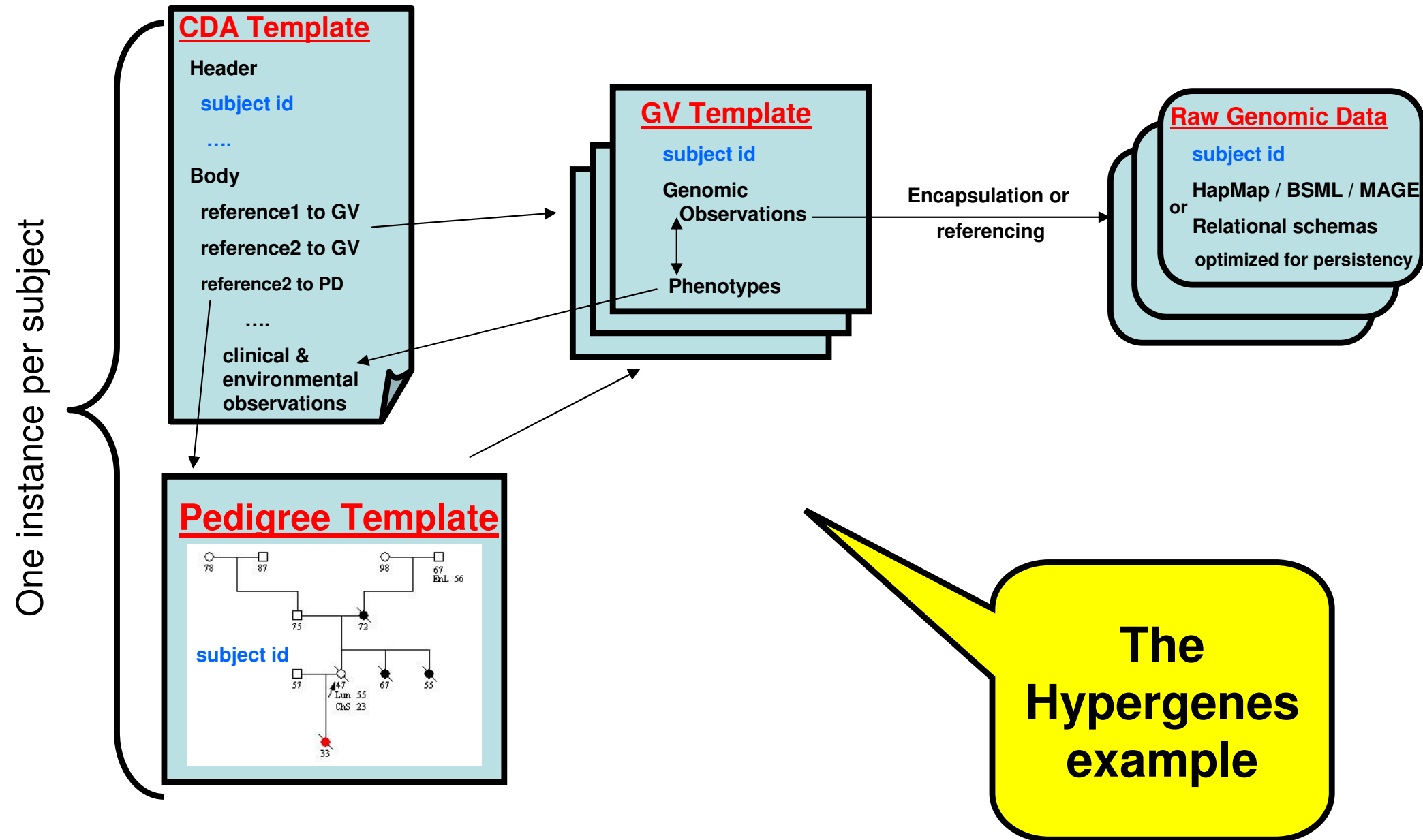
# Hypergenes Work Packages



# The Biomedical Information Infrastructure (BII) Landscape



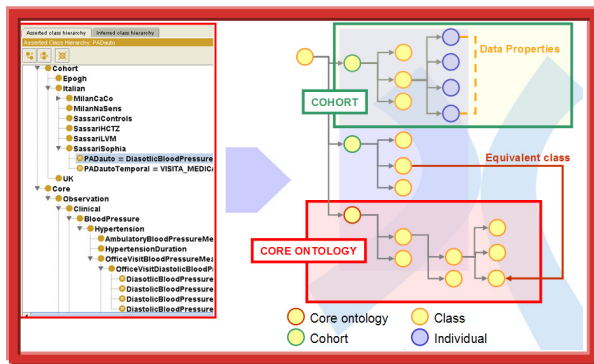
# Information Models over RIMon Warehouse





# Instance Generation (Data & Knowledge)

## OWL Ontology



## Data Source

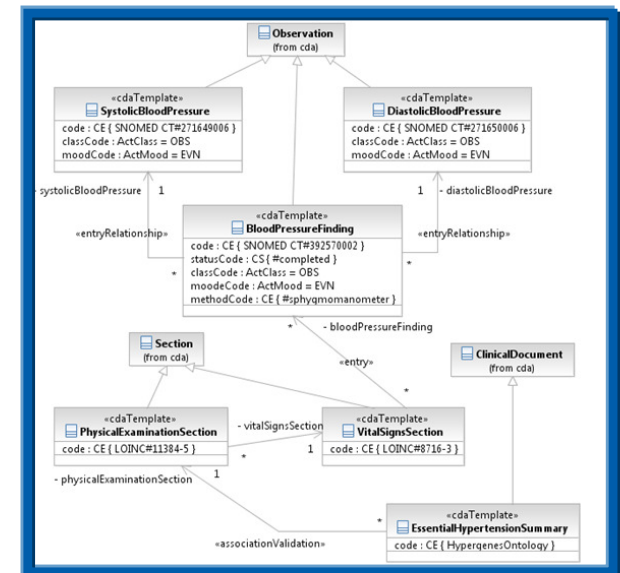
Adapter

## Instance Generation Engine

CTS

Java API

## Template Model



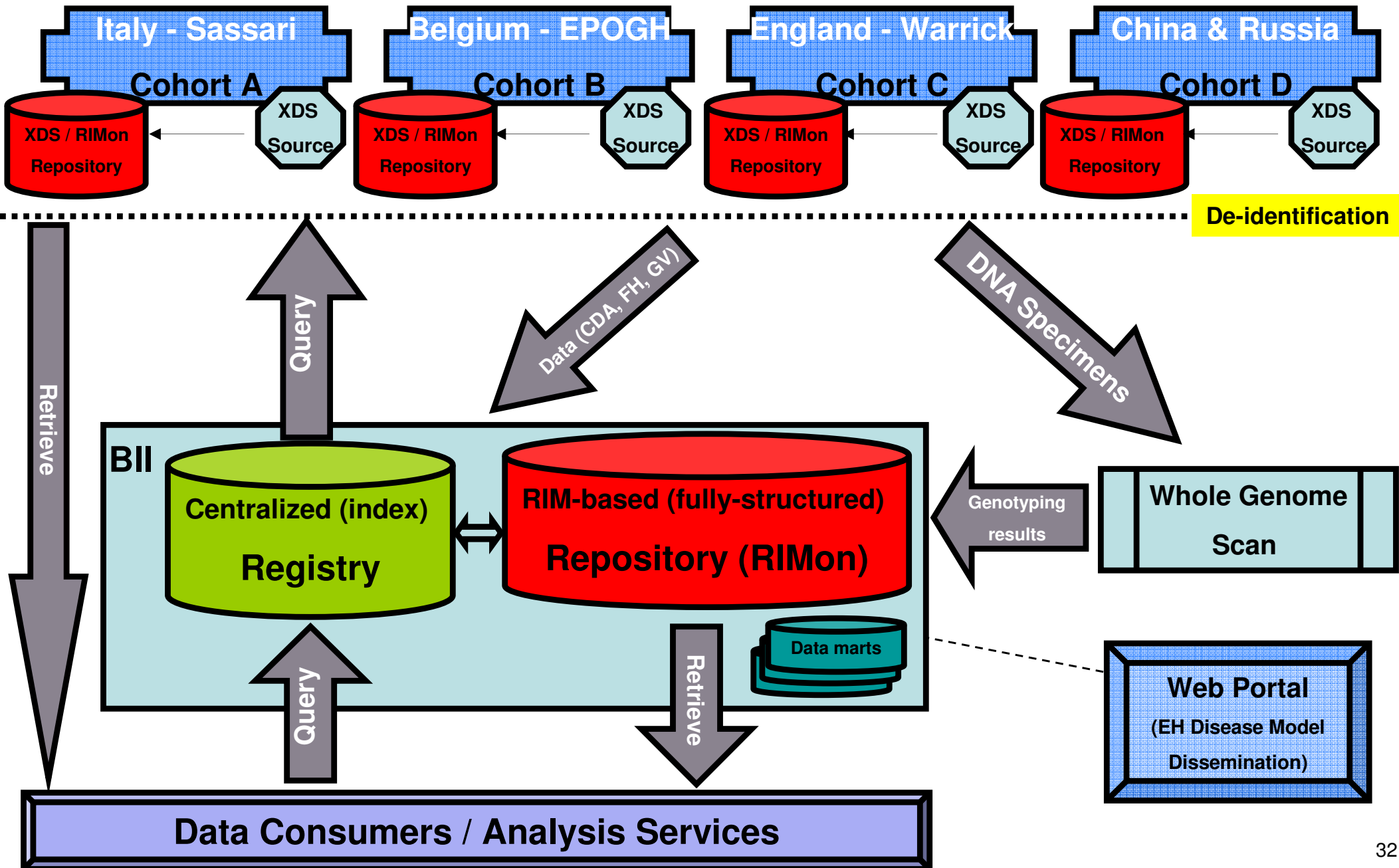
## Representing constraints

## Standard-based Instances (e.g., CDA)

```
<observation classCode="OBS" moodCode="EVN">
  <code code="392570002" codeSystem="2.16.840.1.113883.6.96" codeSystemName="SNOMED CT" displayName="Blood pressure finding">
    <qualifier>
      <name displayName="setting"/>
      <value code="185389009" codeSystem="2.16.840.1.113883.6.96" codeSystemName="SNOMED CT" displayName="Follow-up visit"/>
    </qualifier>
  </code>
  <status code="completed"/>
  <effectiveTime>
    <width unit="week" value="4"/>
  </effectiveTime>
  <methodCode displayName="Sphygmomanometer"/>
  <entryRelationship typeCode="OBS" moodCode="EVN">
    <observation classCode="OBS" moodCode="EVN">
      <code code="271649006" codeSystem="2.16.840.1.113883.6.96" codeSystemName="SNOMED CT" displayName="Systolic BP"/>
      <value unit="mmHg" value="167" xsi:type="PQ"/>
    </observation>
  </entryRelationship>
  <entryRelationship typeCode="OBS" moodCode="EVN">
    <observation classCode="OBS" moodCode="EVN">
      <code code="271650006" codeSystem="2.16.840.1.113883.6.96" codeSystemName="SNOMED CT" displayName="Diastolic BP"/>
      <value unit="mmHg" value="98" xsi:type="PQ"/>
    </observation>
  </entryRelationship>
  <entryRelationship typeCode="OBS" moodCode="EVN">
    <observation classCode="OBS" moodCode="EVN">
      <code code="6797001" codeSystem="2.16.840.1.113883.6.96" codeSystemName="SNOMED CT" displayName="Mean BP"/>
      <value unit="mmHg" value="121" xsi:type="PQ"/>
    </observation>
  </entryRelationship>
</observation>
```

Conform to the  
Template Model

# Potential Support of Distributed Repositories – Extended IHE XDS



Family Health History - Family Health History - Mozilla Firefox

File Edit View History Bookmarks Tools Help

# My Family Health Portrait

A tool from the U.S. Surgeon General

Home Create New History Open Saved History

## Headline for Family History

Some text to describe what to do now that you have your family history report.

Add Another

| Name                                  | Relation |                |
|---------------------------------------|----------|----------------|
| <b>My Family</b>                      |          |                |
| Dan                                   | Self     |                |
|                                       | SON      |                |
|                                       | DAU      |                |
|                                       | NBRO     |                |
|                                       | NSIS     |                |
| <b>My Father's Side of the Family</b> |          |                |
|                                       | PAUNT    |                |
|                                       | PUNCLE   |                |
| <b>My Mother's Side of the Family</b> |          |                |
|                                       | MAUNT    | Maternal Aunt  |
|                                       | MUNCLE   | Maternal Uncle |

Import Family History Save Family History

Opening Dan\_FamilyHistory.xml

You have chosen to open

**Dan\_FamilyHistory.xml**  
which is a: XML Document  
from: https://demo.5amsolutions.com

What should Firefox do with this file?

☐ Open with XML Editor (default)

☒ Save File

☐ Do this automatically for files like this from now on.

OK Cancel

US Surgeon General adopted HL7 Pedigree

Done demo.5amsolutions.com

Visit: <https://familyhistory.hhs.gov/>

# HughesRiskApps complies with the HL7 standard

- Data can be shared with any HL7 compliant software
- Data can be uploaded or downloaded to any EHR that has a complete family history section and that is HL7 compliant
- <http://www.hughesriskapps.net/>

Synthesis Of Risk

Patient Name: **Mary Test** Unit Number: **09240801** Date Of Birth: **01/02/1966**

Synthesis Of Risk

Breast/Ovarian **Colorectal**

Open Pedigree

| Model | Relative Risk | Breast 5 Year | Breast Lifetime |
|-------|---------------|---------------|-----------------|
| Gail  | N/A           | 4.07 %        | 41.8 %          |

Explain

| Model             | Risk of Mutation | Breast 5 Year | Breast Lifetime * | Ovarian 5 Year | Ovarian Lifetime * |
|-------------------|------------------|---------------|-------------------|----------------|--------------------|
| Population Risk   |                  | 0.72 %        | 11.33 %           | 0.07 %         | 1.29 %             |
| Claus             |                  | 1.38 %        | 14.35 %           |                |                    |
| BRCA Pro          | 40.69 %          | 4.82 %        | 27.83 %           | 1.35 %         | 18.88 %            |
| Myriad            | 14 %             | 2.12 %        | 16.95 %           | 0.5 %          | 8.06 %             |
| Synthesis Of Risk | 41 %             | 4.85 %        | 27.95 %           | 1.36 %         | 21.31 %            |

Genetic Testing

Guideline: Consider testing a relative Explain

Clinician: Consider testing a relative

Exit < Back Next >

Slide 4 of 9 Default Design English (U.S.)

start riskAppsV2 Microsoft... riskApp... Main Synthes... untitled ... 34% 3:10 PM



# Biomarker Imaging Management

## BioMIMS – Rizzoli



### ■ The Client

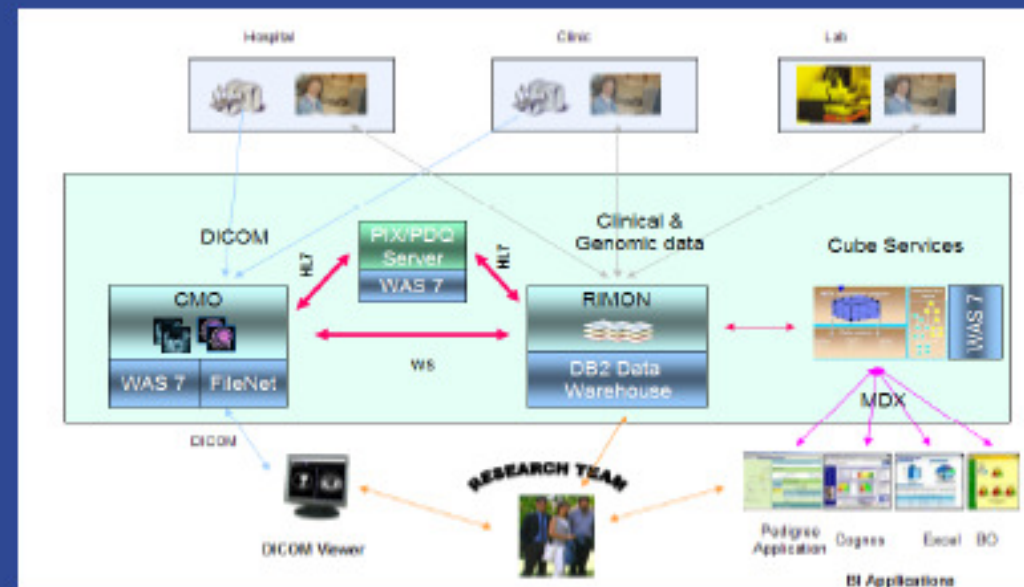
- ▶ Medical Genetic Unit of Istituto Ortopedico Rizzoli (IRO)

### ■ Goal

- ▶ Imaging biomarkers hold tremendous potential for accelerating the development of pharmaceuticals and therapeutic devices, as well as for improving the quality of patient care
- ▶ Develop an imaging biomarkers management solution, leveraging the correlation of bio-medical imaging, clinical and genomic data, based on healthcare standards
- ▶ Support sophisticated analytics and queries, with special emphasis on pedigree visualization

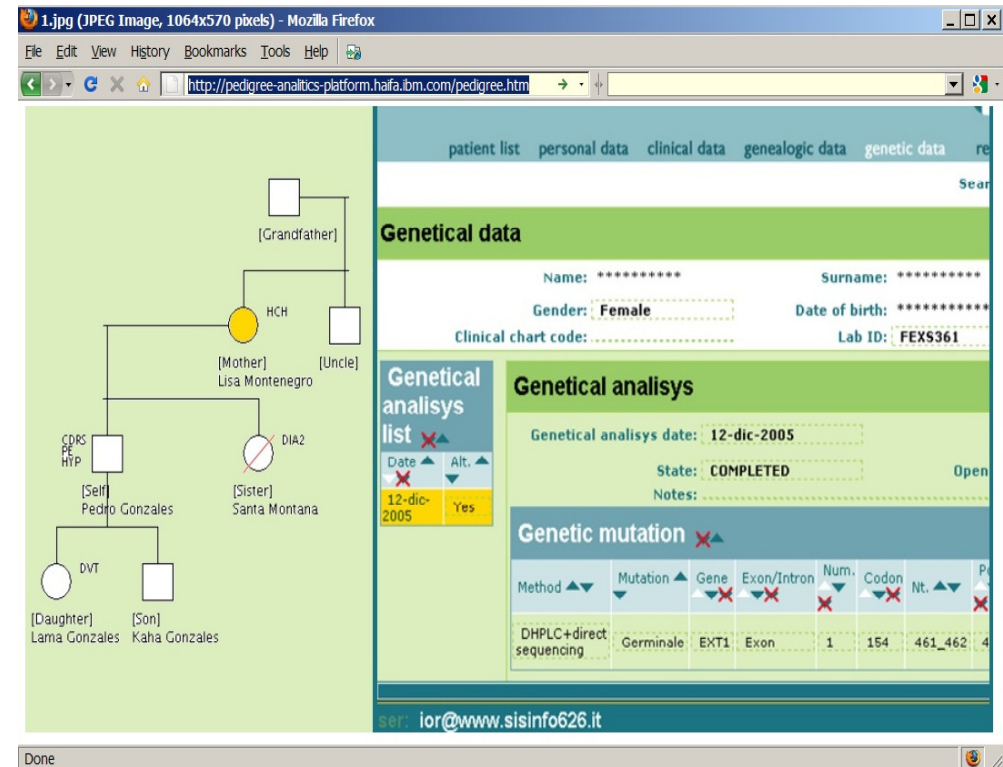
### ■ Challenges

- ▶ Collaborative environment with regional, national and international projects, on skeletal genetic diseases
- ▶ Extensive usage of Imaging data, Genomics data, Clinical Data, Pedigree Information



# BioMIMS - Pedigree Visualization & Access

- Dynamic pedigree visualization
- Presentation of all available information for the persons in the pedigree
  - Clinical, genomic data and medical images
- Standard pedigree representation
  - HL7 v3 Family History
  - Enables standard based pedigree interoperability
- Enables disease risk assessment



## **v2 Implementation Guide (of Lab)**

- **The IG “Genetic Test Result Reporting to EHR” passed informative ballot**
- **It is modeled after the HL7 Version 2.5.1 Implementation Guide: Orders And Observations; Interoperable Laboratory Result Reporting To EHR (US Realm), Release 1**
- **Is used in a pilot of information exchange between Partners Healthcare and Intermountain Health Care**

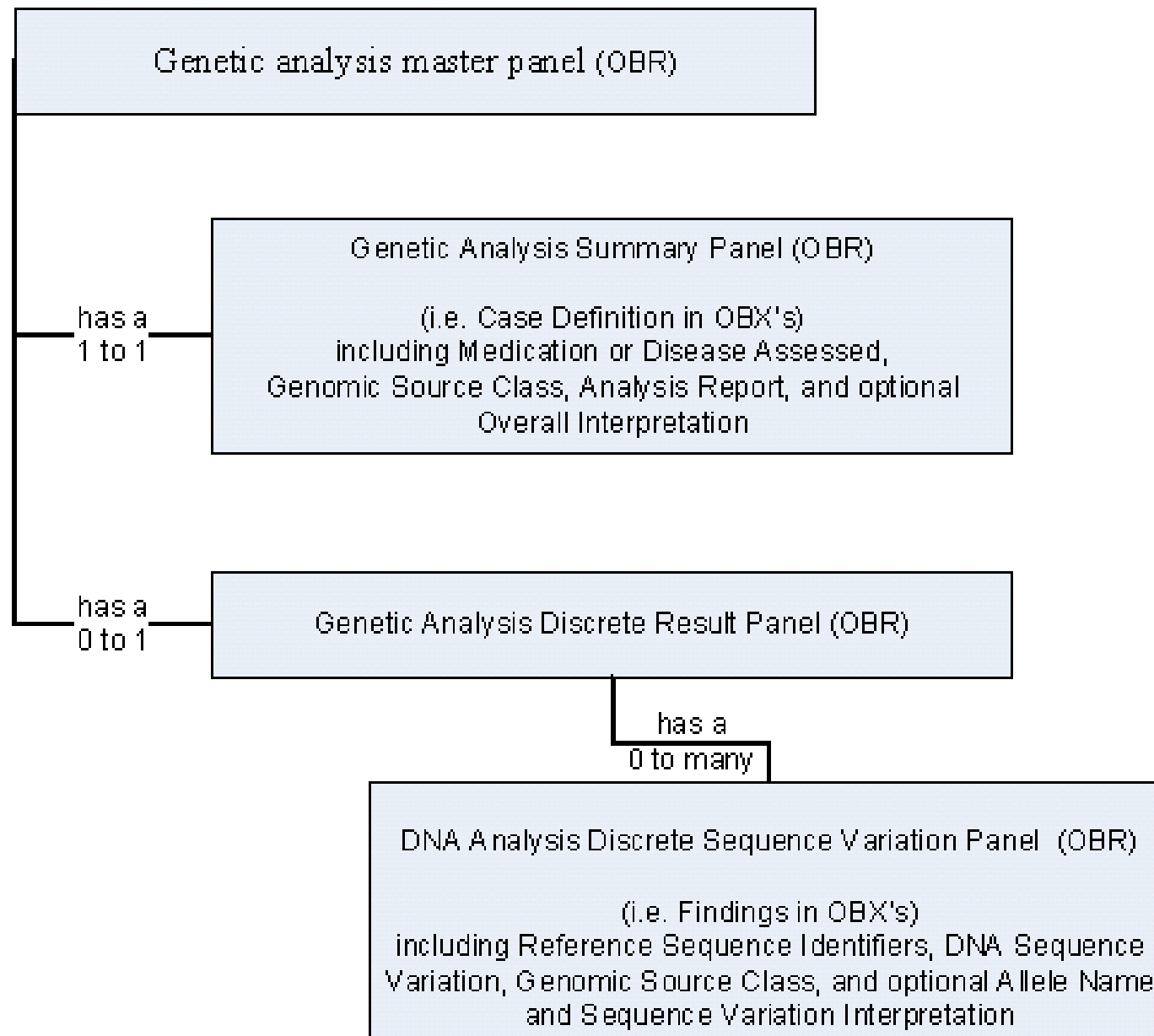


## **v2 Implementation Guide (of Lab)**

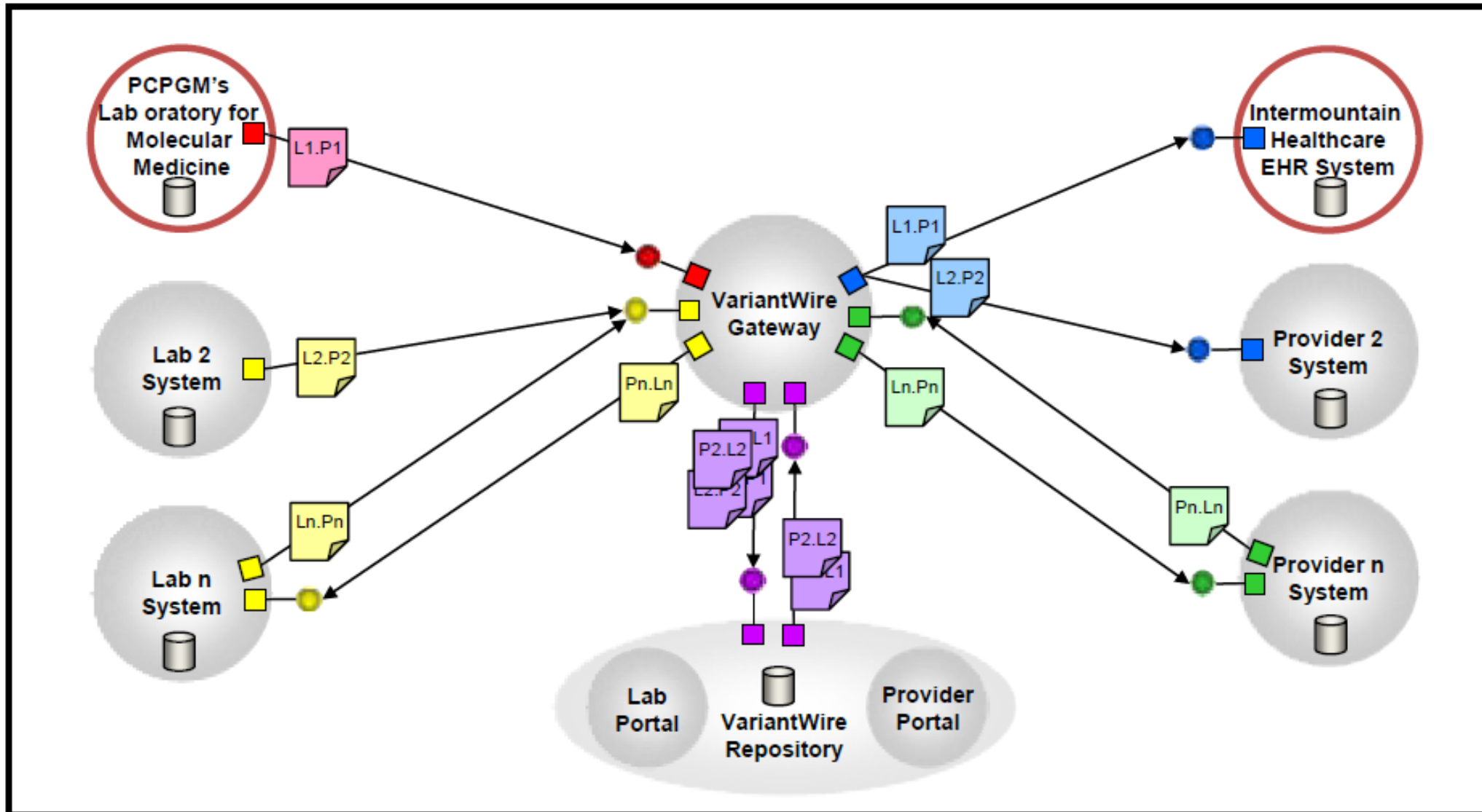
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# The v2 Message Structure



# Harvard – Intermountain Exchange Pilot



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Source: Emerging Clinical Genomics Standards, Mollie Ullman-Cullere, Oct.15, 2009

## v2 Sample Message (Harvard – IHC Pilot)

- OBR|1||PM-08-J00094^HPCGG-LMM^2.16.840.1.113883.3.167.1^ISO||Im\_DCM-pnlB\_L^Dilated Cardiomyopathy Panel B (5 genes)^99LMM-ORDER-TEST-ID||20080702000000|20080702100909||||||234567891^Pump^Patrick^^^^^NPI^L|||||20080703000000||F|||||00000009^Cardiovascular^99HPCGG-GVIE-INDICATION^^^^^Clinical Diagnosis and Family History of DCM|&Geneticist&Gene&&&&NPI^^^^^^HPCGG-LMM&2.16.840.1.113883.3.167.1&ISO|||||||55233-1^Genetic analysis master panel ^LN
- SPM|1|||119273009&Peripheral blood&SNM3&&&&0707Intl&&Blood, Peripheral|||||||20080702000000
- OBR|2||PM-08-J00094-1^HPCGG-LMM^2.16.840.1.113883.3.167.1^ISO|55232-3^Genetic analysis summary panel^LN|||20080702000000|||||||20080703000000||F|||^PM-08-J00094&HPCGG-LMM&2.16.840.1.113883.3.167.1&ISO
- OBX|1|CWE|51967-8^Genetic disease assessed^LN||399020009^DCM-Dilated Cardiomyopathy^SNM3^^^0707Intl|||||F|20080702100909|||||||Laboratory for Molecular Medicine^L^22D1005307^^^CLIA&2.16.840.1.113883.4.7&ISO|1000 Laboratory Lane^Ste. 123^Cambridge^MA^99999^USA^B

# Summary

- **Small group coping with**
  - Various HL7 formats: v3, v2 and CDA
  - Clinical & Research environments
- **Developing component models (CMET) to be used in other HL7 domains**
  - Genetic Variation
  - Gene Expression
- **CDA Genetic Testing Report (GTR)**
  - Bridge from raw data to human readable reports and bubbled-up data
  - Model-driven development of standards (use of MDHT CDA Editor)
- **Call for European Participation...!**
  - An out-of-cycle meeting to kick-off European involvement
  - Collocated with major EU venues (e.g., MIE 2011)



# The End

- Thank you for your attention... 😊
- Questions? Contact Amnon at [shabo@il.ibm.com](mailto:shabo@il.ibm.com)
- Comments of general interest should be posted to the CG mailing list at [clingenomics@lists.hl7.org](mailto:clingenomics@lists.hl7.org)